

Medical Informatics Europe – MIE 2020,
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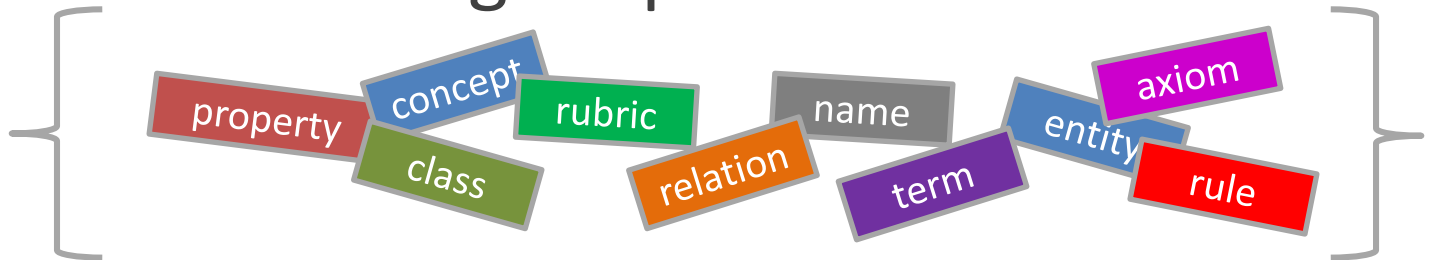
Formal ontologies and multilingual terminologies as tools for knowledge level interoperability in the biomedical domain

Stefan Schulz, Medical University of Graz, Austria

Problem

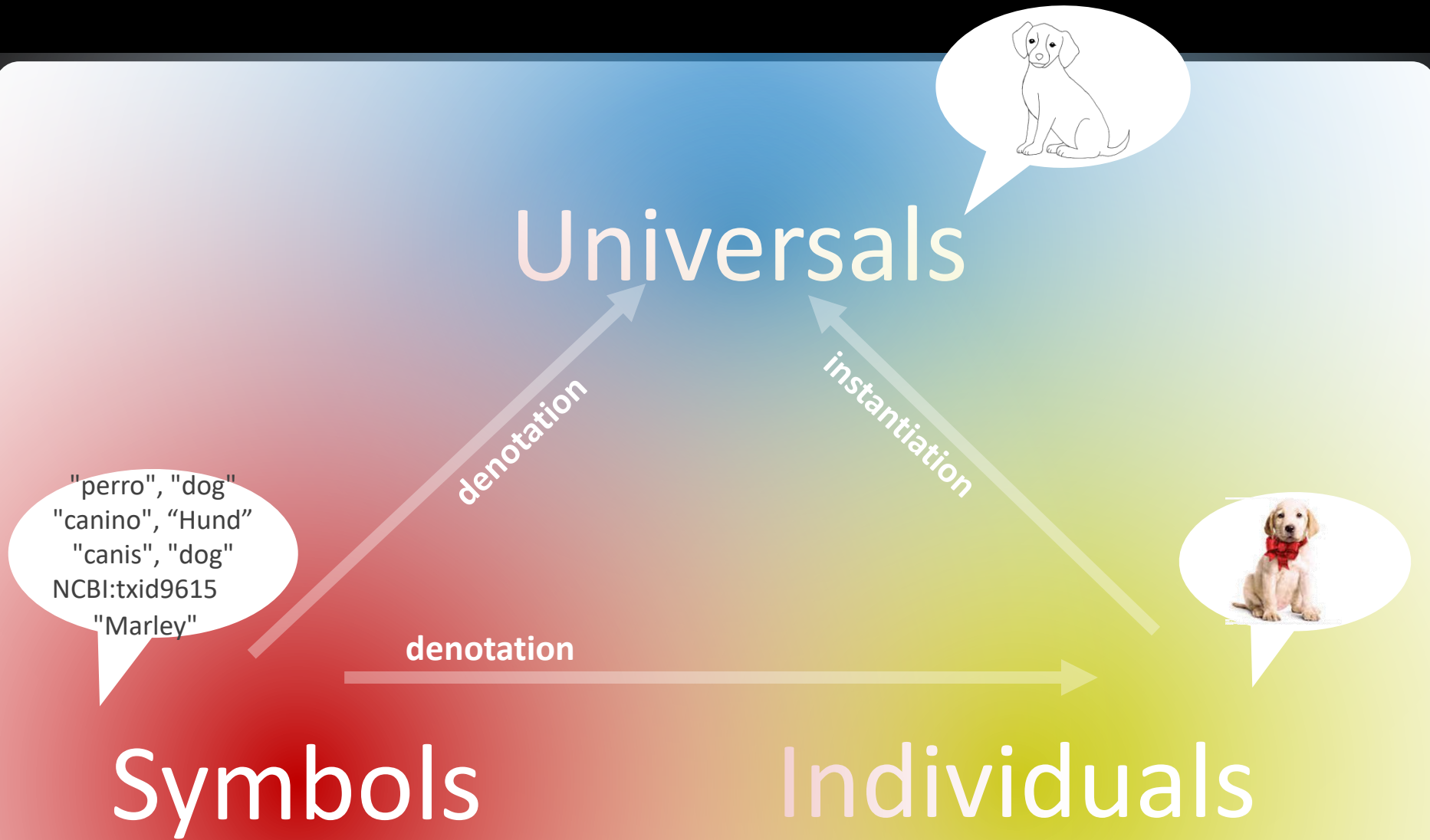
- Babylonian language confusion in biomedical semantics & knowledge representation

Knowledge

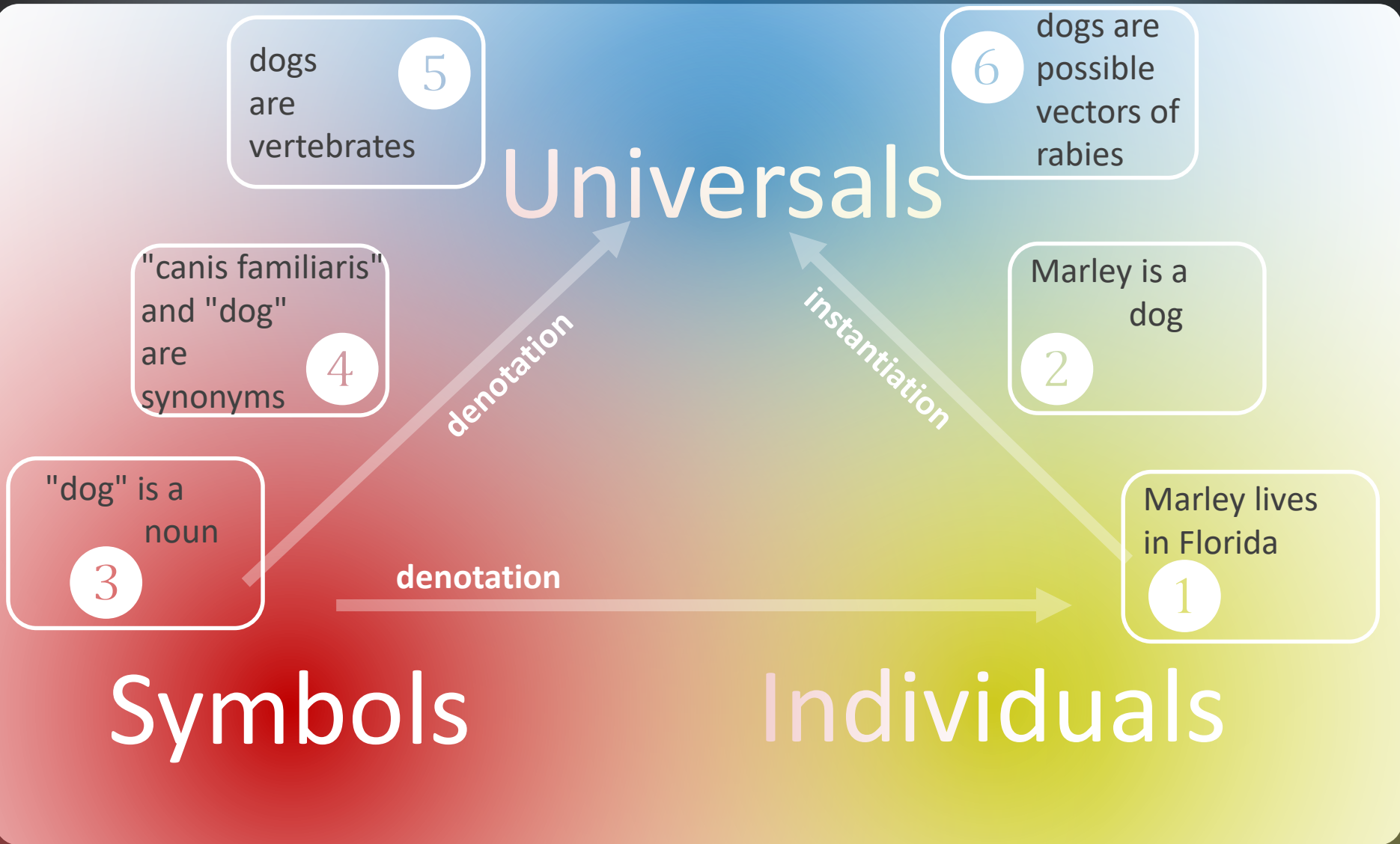


- What kinds of knowledge need to be represented?
- Is a more principled framework possible?
- How do biomedical formal ontologies and multilingual terminologies fit in this picture?

Knowledge map



Knowledge map

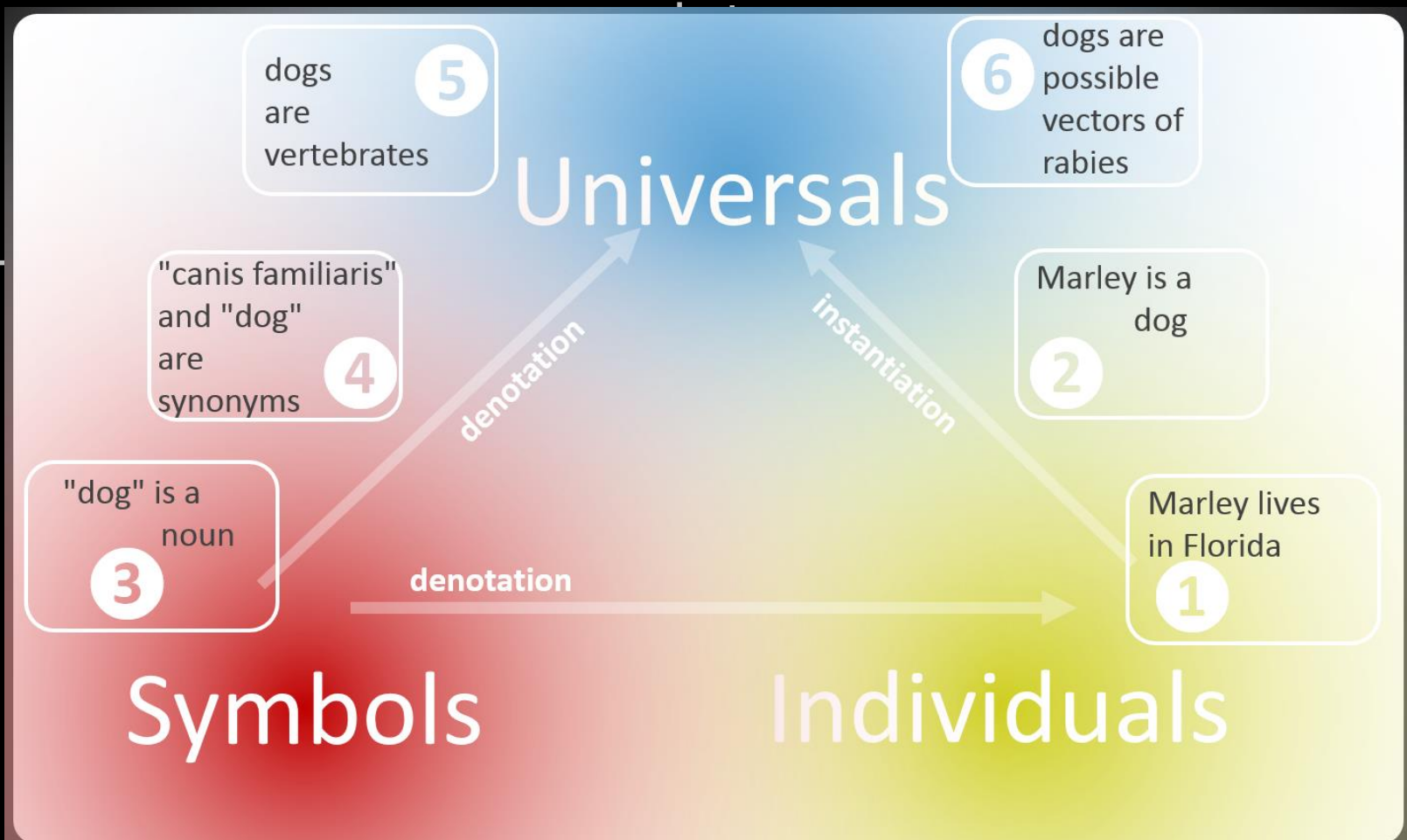


Ontological "knowledge":

Axioms that are universally true

Contingent knowledge:

typical, likely, possible

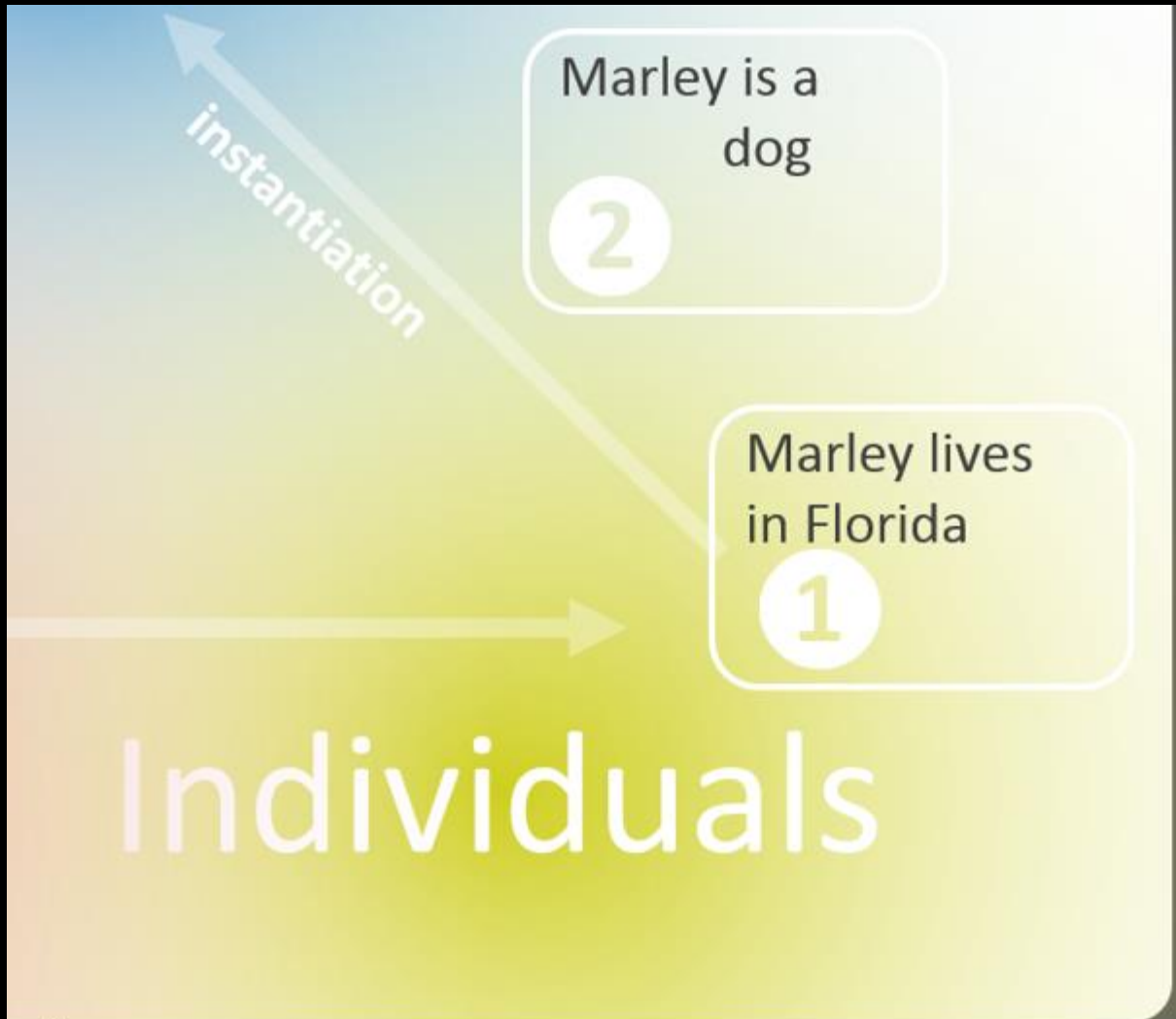


Symbolic knowledge:

Statements about properties and meaning of signs of language

Factual knowledge:

Statements about concrete entities and their relationships



Factual knowledge:

Statements about concrete entities
and their relationships

Statements about individuals

```
<Subject> <Predicate> <Object>  
:Florida rdf:type :state  
:Marley rdf:type :dog  
:Marley :lives :Florida
```

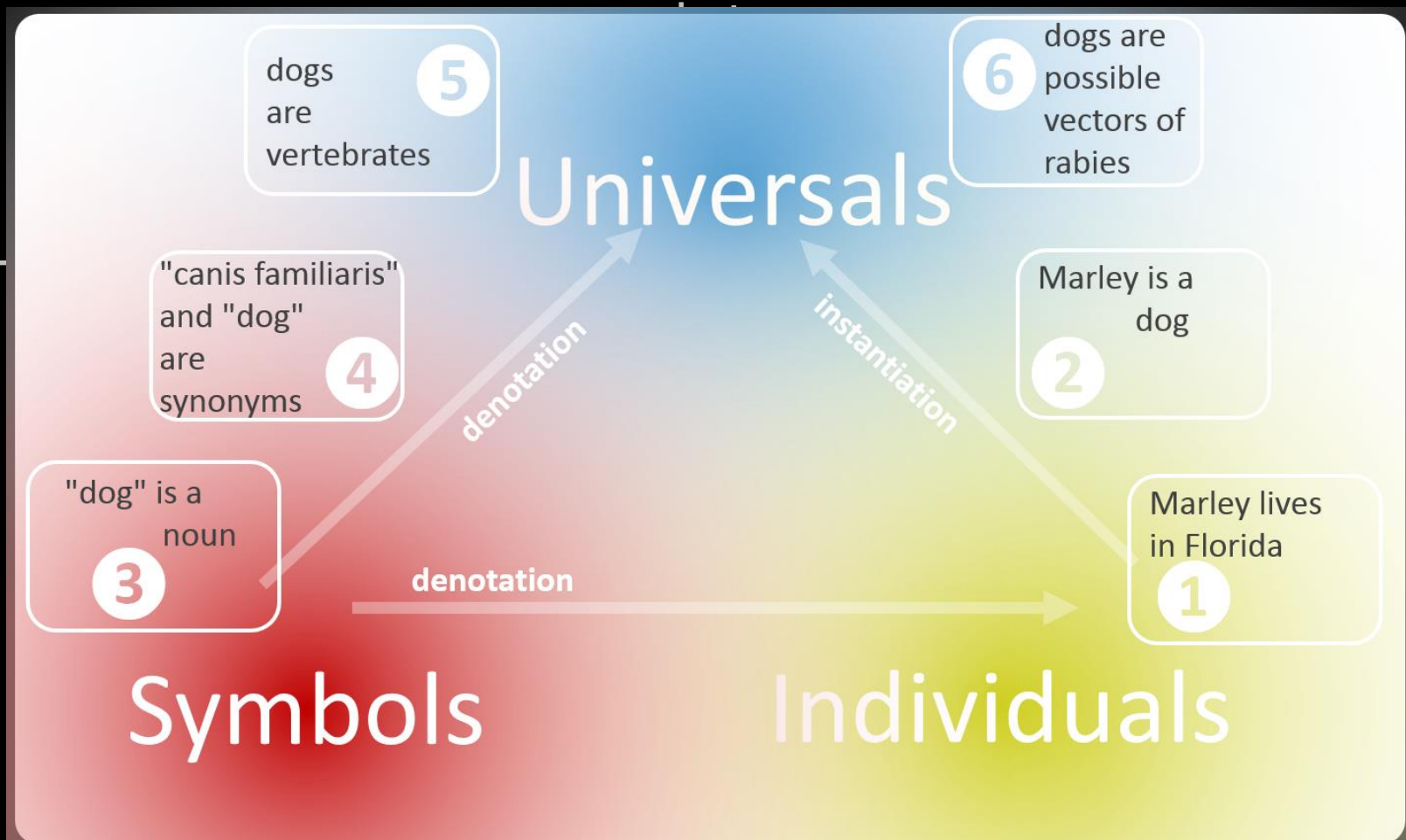
Syntax TURTLE : <https://www.w3.org/TR/turtle/>

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Factual knowledge:

Statements about concrete entities and their relationships

"canis familiaris"
and "dog"
are
synonyms **4**

"dog" is a
noun

3

denotation

denotation

Symbols

Symbolic knowledge

Statements about properties and
meaning of signs of language

Representations SKOS / Linked Data

```
:ex:Dog rdf:type skos:Concept  
:ex:Dog skos:prefLabel "dog"@en;  
:ex:Dog skos:prefLabel "perro"@es;
```

```
:ex:Animal rdf:type skos:Concept  
:ex:Animal skos:broader ex:Dog
```

```
wr:dog      lemon:sense      wr:dog-English-Noun-1  
wr:dog      lemon:sense      wr:dog-English-Verb-1  
wr:dog-English-Noun-1      wt:hasPoS wt:Noun
```

Syntax TURTLE : <https://www.w3.org/TR/turtle/>

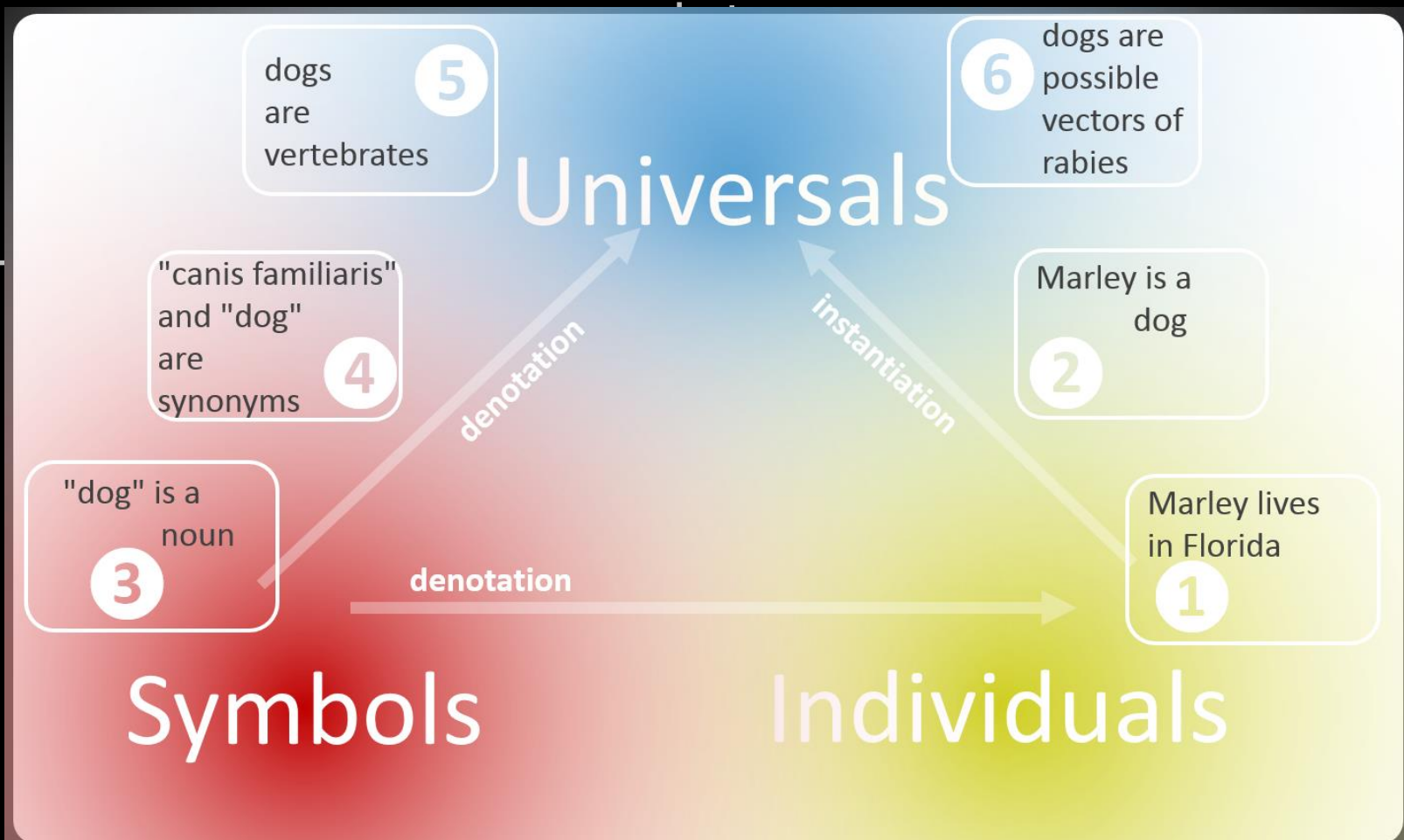
Wiktionary: <http://wiki.dbpedia.org/wiktionary-rdf-extraction>

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Factual knowledge:

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6

Ontological knowledge:

Axioms that are universally true

dogs
are
vertebrates

5

Universals

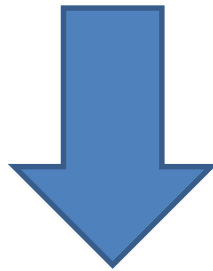
Representation OWL

Dog subclassOf Vertebrate

Vertebrate subclassOf Animal

Vertebra subclassOf Bone

Vertebrate equivalentTo Animal and
has-part some Vertebra



computable inference
(e.g. Hermit or Fact++
OWL reasoner)

There is no dog that has no bones

OWL Manchester Syntax: <https://www.w3.org/TR/owl2-manchester-syntax/>

Hermit reasoner: <http://www.hermit-reasoner.com/>

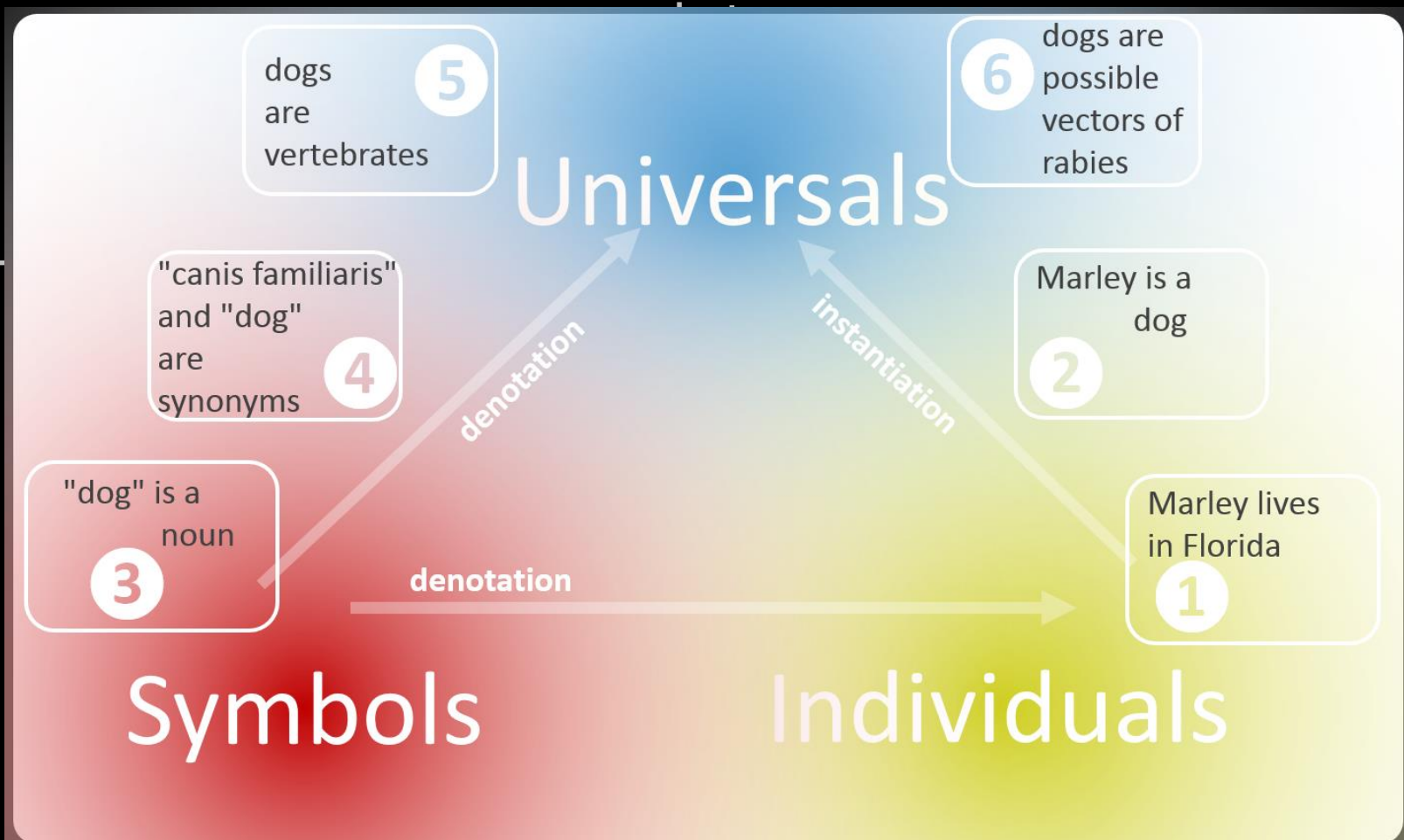
Fact++ reasoner: <http://owl.man.ac.uk/factplusplus/>

Ontological knowledge:

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Contingent knowledge:

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Symbolic knowledge:

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e

Contingent knowledge:
typical, likely, possible

Universals

6

dogs are
possible
vectors of
rabies

Triple representation

- No formal semantics!
- Different, mostly complex interpretations
- Don't use formal languages for this

<Subject>	<Predicate>	<Object>
:Dog	:vector-of	:Rabies
:Tobacco	:causes	:Cancer
:Aspirin	:treats	:Pain
:Fever	:suggests	:Malaria
:Bird	:capable-of	:Flying

~~Dog subclassOf vector-of some Rabies~~

~~Tobacco subclassOf causes some Cancer~~

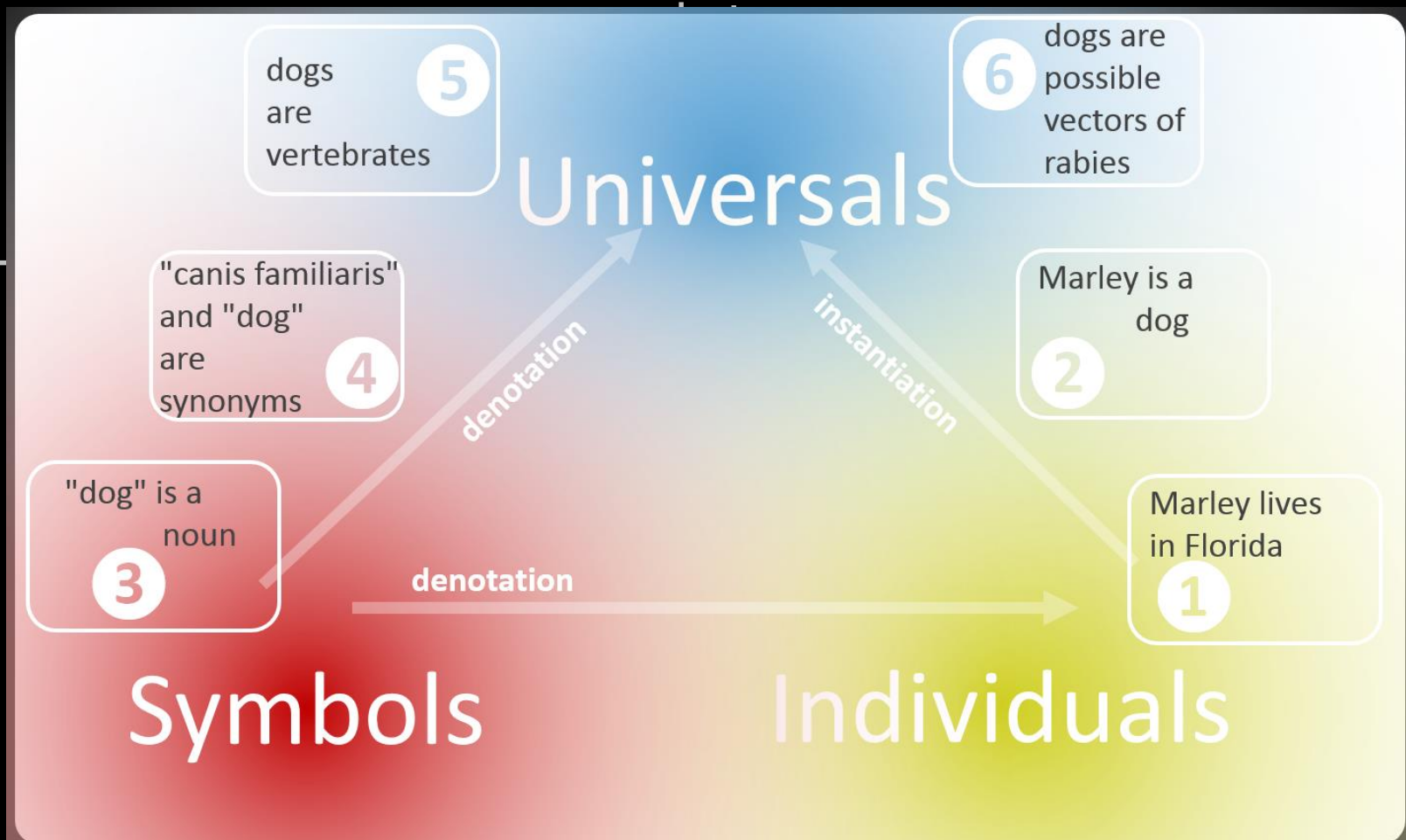
~~Aspirin subclassOf treats some Pain~~

Ontological knowledge:

Axioms that are universally true

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Symbolic knowledge:

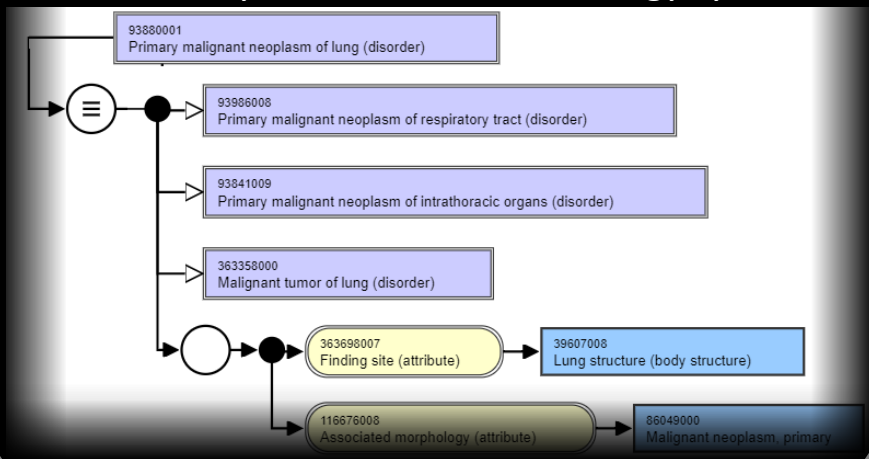
Statements about properties and meaning of signs of language

Factual knowledge:

Statements about concrete entities and their relationships

Ontological knowledge

Axiomatic layer of clinical terminology systems



Contingent knowledge

e.g. Research databases Clinical guidelines

UniProtKB - Q71M42 (PC11X_PANTR)

Protein | **Protocadherin-11 X-linked**
 Gene | **PCDH11X**
 Organism | *Pan troglodytes (Chimpanzee)*

Status | Reviewed - Annotation score: ●●●○○
 - Protein inferred from homology¹

Function¹

potential calcium-dependent cell-adhesion protein. *By similarity*

GO - Molecular function¹

- calcium ion binding *Source: InterPro*

View the complete GO annotation on QuickGO ...

GO - Biological process¹

- cell adhesion *Source: GO_Central*
- homophilic cell adhesion via plasma membrane adhesion molecules
- negative regulation of phosphatase activity *Source: UniProtKB*

>2 symptoms of UTI?

yes → Consider alternative diagnosis

Vaginal discharge or irritation? yes → Consider pelvic exam and further investigations

no

Back/flank pain or fever? yes → **Consider upper UTI**

no

Risk factors for complicated UTI? yes → Dipstick urine, Consider urine C+S

no → **Diagnose lower UTI**

No need for urine dipstick

No need for urine C+S

Consider empirical treatment

Consider empirical therapy for 7 days

Review in 24 hours

If no response post 24 hours consider admission



Primary malignant neoplasm of lung (disorder)

SCTID: 93880001

en Primary malignant neoplasm of lung	da Primært malignt neoplasme i lunge
en Lung cancer	sv primär malign tumör i lunga
en Primary malignant neoplasm of lung (disorder)	nl primair maligne neoplasma van lung
es neoplasia maligna primaria de pulmón	fr tumeur maligne primaire du poumon

Lexical layer of terminology systems

Symbolic knowledge



e.g. Clinical Information Models

```

Condition (DomainResource)
identifier : Identifier [0..*]
clinicalStatus : CodeableConcept [0..1] «
    ConditionClinicalStatusCodes! »
verificationStatus : CodeableConcept [0..1] «
    ConditionVerificationStatus! »
category : CodeableConcept [0..*] « ConditionCategoryCodes+ »
severity : CodeableConcept [0..1] « Condition/DiagnosisSeverity? »
code : CodeableConcept [0..1] « Condition/Problem/DiagnosisCo...?? »
bodySite : CodeableConcept [0..*] « SNOMEDCTBodyStructures?? »
subject : Reference [1..1] « Patient|Group »
encounter : Reference [0..1] « Encounter »
onset[x] : Type [0..1] « dateTime|Age|Period|Range|string »
abatement[x] : Type [0..1] « dateTime|Age|Period|Range|string »
recordedDate : dateTime [0..1]
recorder : Reference [0..1] « Practitioner|PractitionerRole|Patient
    
```

Factual knowledge